

Multicentric Non-monomelic Synchronous Chondrosarcoma Secondary to Maffucci Syndrome: Approach to Diagnosis and Treatment

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Abstract

Maffucci syndrome presents with combination of enchondromas and hemangiomas with rare sarcomatous transformations of the lesions. We present a rare case of such sarcomatous transformation in a 64 year old male who presented with ankle swelling since one year. MRI showed a large soft tissue mass having lobular swelling with irregular calcification within the mass in very low signal areas and endosteal scallopings, which was suggestive of a possibility of malignant transformation. PET Scan revealed a destructive lesion at ankle along with another lesion in coracoid process of scapula. Biopsy showed well differentiated chondrosarcoma and hence a diagnosis of multicentric non-monomelic synchronous chondrosarcoma secondary to maffucci syndrome was made. The case is rare in terms of having sarcomatous transformation in two different lesions

Keywords: Maffucci syndrome, sarcomatous transformation, multicentric

Introduction

Maffucci's syndrome is rare, congenital, non hereditary mesodermal dysplasia clinically presents as enchondromas and hemangiomas. Maffucci syndrome was first described in 1881 and more than 200 cases are reported since then [1]. It has unknown etiology and no predilection for sex or race. Enchondromas are benign cartilaginous tumors most commonly seen in phalanges and long bones. Hemangiomas are most frequently located in the dermis and subcutaneous fat adjacent to areas of enchondromatosis. The development of lesions usually occurs in puberty. It is associated with secondary musculoskeletal deformities and rarely sarcomatous transformation. Complications are fracture, sarcomatous

degeneration or chondrosarcomatous transformation. The risk of spontaneous fracture through area of advanced rarefaction in 25-30% and sarcomatous transformation is about 15-30%. Chondrosarcomatous transformation occurs in approximately 40-50% of enchondromas. We present a rare case of non monomelic synchronous development of chondrosarcoma in a case of Maffucci syndrome. We have stressed on the importance of careful surveillance for malignant degeneration of both skeletal and non-skeletal lesions.

Case Report

A 64-year-old male presented with pain and swelling of right ankle (fig.1) which was insidious in onset and progressive in nature from last 1 year. Patient also had difficulty in standing and walking. On examination patient had soft to firm swelling of about 10 cms in diameter with no signs of inflammation. Swelling was tender, soft to firm and non-mobile on the right mid-foot region. In addition, complete physical

examination revealed multiple soft tissue swellings of right hand (Fig.2&3.) with multiple deformities of right upper (Fig.4) and lower limbs since birth. Patient was able to all activities of daily living. He was investigated in the form of radiographs which revealed (Fig.5&6) fusiform, lytic lesion, intralesional calcifications, endosteal scallopings and occasional periosteal reaction in right foot. Radiograph of (Fig.7) right hand showed multiloculated, septate, lytic, well-defined, irregularly expanded and radiolucent lesion with intact cortex suggesting enchondromas and moreover multiple soft tissue swellings containing speckled calcifications suggesting hemangiomas with phlebolith over right hand. To investigate further MRI of right ankle (Fig.8 &9) was done that showed high signal intensity on T2 sequences and low intensity on T1 sequences, and large soft tissue mass having lobular swelling with irregular calcification within the mass in very low signal areas and endosteal scallopings, which was suggestive of a possibility of malignant

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Figure 1: 64-year-old male presented with pain and swelling of right ankle

Figure 2: multiple soft tissue swellings of right hand (a,b) with multiple deformities of right upper (c)

transformation. A PET-SCAN was done in order to evaluate all the lesions and to rule out multicentric disease. PET SCAN Revealed a (fig.10) hypermetabolic destructive, osteolytic lesion in right foot with large soft tissue component involving tarsal bone and

size of soft tissue is 8.9cm x 6.5cm x 6.3cm. In addition, it also showed a new FDG avidin corocoid process of right scapula. Radiograph (fig 11) and MRI (fig.12)of the right scapula showed lytic lesion with cortical erosion in the coracoid process with small punctuate

area with calcification with cortical erosions of corocoid process of right scapula.

Biopsy from both the sites (right ankle and corocoid process of the right scapula) showed Greyish-white, soft

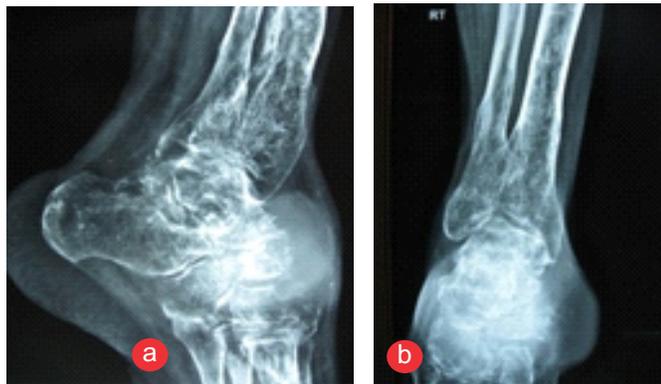


Figure 3 a,b: radiographs which revealed fusiform ,lytic lesion, intralesional calcifications, endosteal scallopings in right foot



Figure 4: Radiograph of right hand showed multiloculated, septate, lytic, well-defined, irregularly expanded and radiolucent lesion with intact cortex suggesting enchondromas and moreover multiple soft tissue swellings containing speckled calcifications suggesting hemangiomas

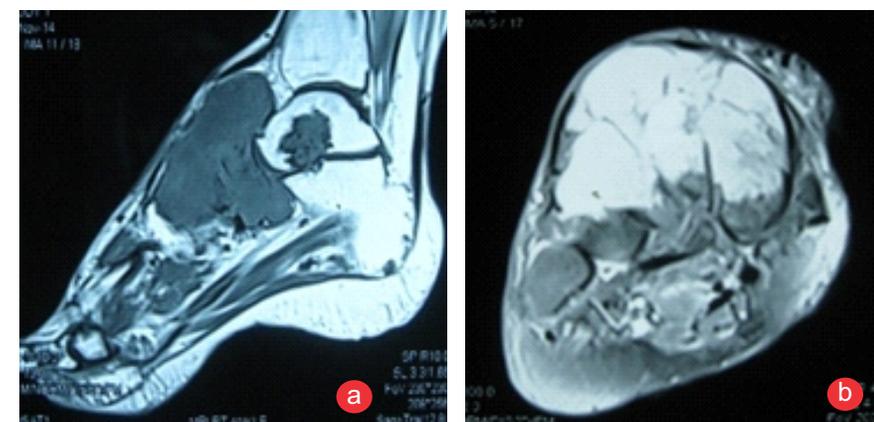


Figure 5 a,b: MRI showed high signal intensity on T2 sequences and low intensity on T1 sequences, and large soft tissue mass having lobular swelling with irregular calcification within the mass in very low signal areas and endosteal scallopings, which was suggestive of a possibility of malignant transformation

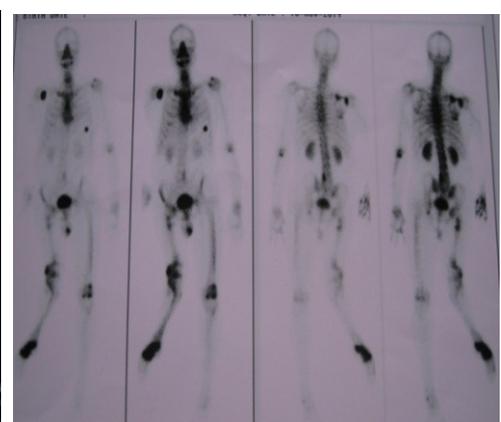


Figure 6: PET scan revealed hypermetabolic destructive, osteolytic lesion in right foot a new FDG avid lesion in corocoid process of right scapula.

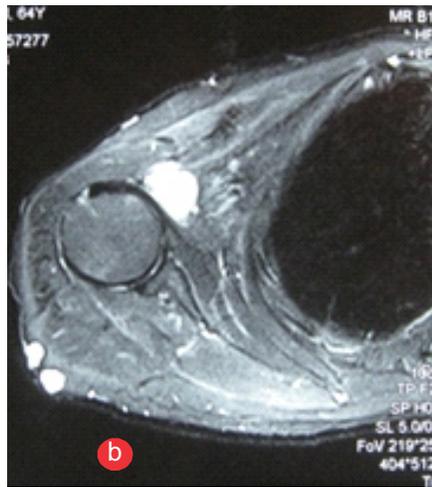


Figure 7 a,b: Radiograph and MRI of the right scapula showed lytic lesion with cortical erosion in the coracoid process with small punctuate area with calcification with cortical erosions of coracoid process of right scapula.

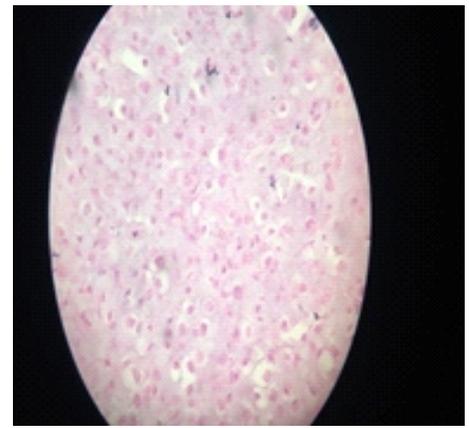


Figure 8: showed multiple fragments composed of chondromyxoid stroma with scattered cells having ground glass appearance, small to medium sized hyperchromatic nuclei with moderate to abundant eosinophilic cytoplasm suggestive of well differentiated chondrosarcoma

tissue from both the sites and microscopy (fig13) showed multiple fragments composed of chondromyxoid stroma with scattered cells having ground glass appearance, small to medium sized hyperchromatic nuclei with moderate to abundant eosinophilic cytoplasm suggestive of well differentiated chondrosarcoma and hence a diagnosis of multicentric non-monomelic synchronous chondrosarcoma secondary to Maffucci syndrome was made. Patient underwent below knee amputation of right lower limb and limb salvage surgery of total scapulectomy right shoulder. Patient was under follow-up and is disease free since then.

Discussion

The diagnosis of Maffucci's syndrome is suggested by the presence of multiple enchondromas and multiple haemangiomas, other conditions in the differential diagnosis include Klippel-Trenaunay syndrome, Sturge-Weber syndrome, von Hippel-Lindau syndrome, and Ollier's disease. Ollier's disease, on the other hand, is only rarely associated with sarcomatous

degeneration of the enchondromas or other systemic neoplasms. This has always been considered as a key difference between Ollier's disease and Maffucci syndrome. Rate of malignant transformation in Maffucci syndrome is 40-50% and in Ollier's disease is 20-25%. Multiple enchondromatosis can usually be diagnosed by clinical and radiological findings; sometimes pathological confirmation is required. Clinically it manifests as gross bony deformities due to intraosseous metaphyseal and diaphyseal cartilaginous masses. The lesions are usually painless, unless associated with pathological fracture or superimposed trauma. Radiologically, the lesions appear as rounded or oval, centrally located intraosseous lucencies commonly associated with matrix calcifications. They may symmetrically or asymmetrically expand the bone. Small bones of the hands and feet are most common sites followed by long bones of the upper and lower limbs. PET-CT interpretations are helpful for diagnosing malignant transformation. SUV >2.0 shows malignant transformation. PET-CT is important test with accuracy of 94% for identifying chondrosarcomas and their

recurrence after surgery. As PET-CT detects hypermetabolic foci, whole body PET-CT should be recommended. PET-CT is also helpful in evaluating the response of sarcomas to neoadjuvant treatment and best responders show greater decline in SUV levels as compared to poor responders. PET-CT also detects lymph nodes and metastases in patients with sarcomas. Malignant transformation of the enchondroma into chondrosarcomas, osteosarcomas and fibrosarcoma is a well known complication in Maffucci syndrome. Individuals having Maffucci syndrome need to have regular physical examination in order to evaluate changes in the skin and bone lesions that may suggest malignancy. Haemangiomas could be treated with sclerotherapy, surgery or with laser treatment to reduce the size of the lesions. Biopsy can be done in case of symptomatic enchondromas. Surgery can be done in case of borderline or low grade chondrosarcoma. Because of low grade of the tumor additional treatment radiotherapy/chemotherapy is not necessary (Birsasteanu F et al) Bone and soft tissue tumors of the feet are uncommon. Most bone tumors are

chondrogenic, but differential diagnosis of malignant from benign disease is difficult and prognosis is poor. Management of benign tumors by marginal resection has good prognosis, whereas prognosis of soft tissue sarcomas is very poor (G Kinoshita et al). Prognosis depends upon severity of the disease but there is an increased risk for malignancy. The rarity in this case was appearance of malignant transformation

at two sites in different limbs at the same time secondary to Maffucci Syndrome

Conclusions

Patient with multiple enchondromas on x-ray, should be examined carefully to determine recurrence. Treatment of Maffucci syndrome should be in the form of early detection of malignant transformation as well as symptomatic relief.

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